

# **Parent Fact Sheet**

## **Disorder**

### **Trifunctional Protein Deficiency**

#### **Cause**

People with trifunctional protein (TFP) deficiency have problems breaking down fat into energy for their body. TFP deficiency occurs when a group of enzymes, called “trifunctional protein” is either missing or not working properly. The job of TFP is to breakdown certain fats from the food we eat into energy. It also breaks down fat already stored in the body.

Energy from fat keeps us going whenever our bodies run out of the main source of energy glucose. Our bodies rely on fat when we do not eat for a stretch of time-like sleeping during the night or missing a meal. When TFP is missing or not working well, the body cannot use fats for energy. Once all the glucose is used up, the body tries to use fat without success. This leads to low blood sugar and to the buildup of harmful substances in the blood.

#### **If Not Treated**

The symptoms can vary from person to person. Babies with early TFP deficiency have episodes of illness called metabolic crisis. Some of the first symptoms are: extreme sleepiness, behavior changes, irritable mood, muscle weakness, poor appetite. Other symptoms may include fever, nausea, diarrhea, vomiting, hypoglycemia, increased levels of acidic substances in the blood, called metabolic acidosis. If a metabolic crisis is not treated, a child with TFP deficiency can develop: breathing problems, seizures, coma-sometimes leading to death.

#### **Treatment Options**

Your doctor will work with a metabolic specialist and dietitian to care for your child. Lifelong treatment is usually needed.

Avoid going a long time without food. These babies need to eat more often to avoid low blood sugar. They should not go without eating for more than 4-6 hours. Some babies will need to eat more often than this. A low fat, high carbohydrate diet is often recommended. Your dietitian will help plan any diet changes.

Medium Chain Triglyceride oil (MCT) and L-Carnitine is often used as part of the food plan for people with TFP deficiency. This special oil has medium chain fatty acids that can be used in small amounts for energy. Your dietitian will tell you how to use this supplement. You will have to get a prescription from the doctor for MCT oil.

#### **If Treated**

For more information go to the following website: <http://www.newbornscreening.info>